

**Amendments to the Claims**

Please cancel Claims 3-8, 12, 15, 19-30, 31 and 33-36 without prejudice. Applicants reserve the right to prosecute such claims or the subject matter thereof in further applications. Please amend claims 1 and 32. Please add new claims 40-52. The Claim Listing below will replace all prior versions of the claims in the application:

**Claim Listing**

1. (Currently amended) An isolated human PAB II gene comprising a polymorphic GCG repeat in exon I thereof, wherein said polymorphic GCG repeat has the sequence  

$$\text{ATG (GCG)}_{6+n} \text{GCA},$$
~~with n being selected from 1 to 7 and wherein said polymorphic repeat of said GCG repeat is indicative of a disease in a human patient, wherein n is selected from the group consisting of:~~
  - a) n=0, wherein said gene is associated with a non-disease phenotype; and
  - b) n is selected from 1 to 7, wherein said gene is associated with a phenotype of oculopharyngeal muscular dystrophy, selected from at least one of protein accumulation in a cell nucleus, swallowing difficulty, and ptosis, andwherein said human PAB II gene comprises the sequence as set forth in SEQ ID NO: 18, excluding nucleotide positions 1283 to 1306 thereof, said nucleotide positions 1283 to 1306 being replaced by said  $\text{ATG (GCG)}_{6+n} \text{GCA}$  sequence.
- 2-8. (Canceled)
9. (Previously presented) An isolated nucleic acid sequence comprising a polymorphic GCG repeat of exon I of a human PAB II gene, wherein said polymorphic GCG repeat has the sequence  

$$\text{ATG (GCG)}_{6+n} \text{GCA},$$
with n being selected from 1 to 7 and wherein said polymorphic repeat of said GCG repeat in a patient's human PAB II gene is indicative of a disease in said human patient.
10. (Canceled)
11. (Previously presented) The nucleic acid sequence of claim 9, wherein n is selected from 2 to 7, and wherein said polymorphic repeat of said GCG repeat is associated with an increased severity of said disease.
12. (Canceled)
13. (Previously presented) A method for the diagnosis or prognosis of oculopharyngeal muscular

dystrophy (OPMD), a disease associated with protein accumulation in a cell nucleus, and/or swallowing difficulty and/or ptosis in a human patient, which comprises:

- a) obtaining a nucleic acid sample of said patient; and
- b) determining allelic variants of a GCG repeat in exon I of the PAB II gene, said GCG repeat having the sequence  

$$\text{ATG (GCG)}_{6+n} \text{GCA},$$

wherein n is selected from 0 to 7, and

whereby at least one of the two alleles of said GCG repeat having an n equal to 1 to 7, is indicative of OPMD.

- 14. (Original) The method of claim 13, wherein n is selected from 2 to 7, and wherein said allelic variant is associated with an increased severity of said disease.
- 15. (Canceled)
- 16. (Previously presented) The method of claim 13, wherein said first allele of said GCG repeat has an n which is equal to 1.
- 17. (Previously presented) The method of claim 16, wherein said second allele of said GCG repeat has an n selected from 2 to 7, and wherein said first allele is a modulator of the severity of the phenotype associated with said second allele.
- 18-31. (Canceled)
- 32. (Currently amended) The human PAB II gene of claim 31, wherein n=0, and wherein the sequence of said gene is as set forth in SEQ ID NO:18.
- 33-36. (Canceled)
- 37. (Previously presented) An isolated PAB II nucleic acid sequence comprising a polymorphic GCG repeat having the sequence

$$\text{ATG (GCG)}_{6+n} \text{GCA},$$

wherein n is selected from the group consisting of:

- a) n=0, wherein said nucleic acid sequence is associated with a non-disease phenotype; and
  - b) n is selected from 1 to 7, wherein said nucleic acid sequence is associated with a phenotype of oculopharyngeal muscular dystrophy, selected from at least one of protein accumulation in a cell nucleus, swallowing difficulty, and ptosis.
- 38. (Previously presented) The isolated nucleic acid sequence of claim 37, wherein n=0, and

wherein said sequence comprises the sequence as set forth in SEQ ID NO:18.

39. (Previously presented) The isolated nucleic acid sequence of claim 37, wherein  $n=0$ , and wherein said sequence comprises the sequence as set forth in SEQ ID NO:1.
40. (New) The isolated nucleic acid sequence of claim 37, wherein  $n=0$ , and wherein said GCG repeat has the sequence set forth in SEQ ID NO:2.
41. (New) The isolated nucleic acid sequence of claim 37, wherein  $n=1$ , and wherein said GCG repeat has the sequence set forth in SEQ ID NO:3.
42. (New) The isolated nucleic acid sequence of claim 37, wherein  $n=2$ , and wherein said GCG repeat has the sequence set forth in SEQ ID NO:4.
43. (New) The isolated nucleic acid sequence of claim 37, wherein  $n=3$ , and wherein said GCG repeat has the sequence set forth in SEQ ID NO:5.
44. (New) The isolated nucleic acid sequence of claim 37, wherein  $n=4$ , and wherein said GCG repeat has the sequence set forth in SEQ ID NO:6.
45. (New) The isolated nucleic acid sequence of claim 37, wherein  $n=5$ , and wherein said GCG repeat has the sequence set forth in SEQ ID NO:7.
46. (New) The isolated nucleic acid sequence of claim 37, wherein  $n=6$ , and wherein said GCG repeat has the sequence set forth in SEQ ID NO:8.
47. (New) The isolated nucleic acid sequence of claim 37, wherein  $n=7$ , and wherein said GCG repeat has the sequence set forth in SEQ ID NO:9.
48. (New) A method for the diagnosis or prognosis of oculopharyngeal muscular dystrophy (OPMD), a disease associated with protein accumulation in a cell nucleus, and/or swallowing difficulty and/or ptosis in a human patient, which comprises:
  - a) determining allelic variants of a GCG repeat in exon I of the PAB II gene from a sample of a patient, said GCG repeat having the sequence  
ATG (GCG)<sub>6+n</sub> GCA,wherein  $n$  is selected from 0 to 7, and  
whereby at least one of the two alleles of said GCG repeat having an  $n$  equal to 1 to 7, and is indicative of OPMD.
49. (New) The method of claim 48, wherein  $n=0$ , and wherein said GCG repeat has the sequence set forth in SEQ ID NO:2.
50. (New) The method of claim 48, wherein said first allele of said GCG repeat has an  $n$  which is equal to 1, and has the sequence set forth in SEQ ID NO:3.

51. (New) The method of claim 48, wherein  $n$  is selected from 2 to 7, wherein said allelic variant is associated with an increased severity of said disease, and wherein said GCG repeat has the sequence selected from the group consisting of:
- a) SEQ ID NO:4, when  $n=2$ ;
  - b) SEQ ID NO:5, when  $n=3$ ;
  - c) SEQ ID NO:6, when  $n=4$ ;
  - d) SEQ ID NO:7, when  $n=5$ ;
  - e) SEQ ID NO:8, when  $n=6$ ; and
  - f) SEQ ID NO:9, when  $n=7$ .
52. (New) The method of claim 50, wherein said second allele of said GCG repeat has an  $n$  selected from 2 to 7, said first allele is a modulator of the severity of the phenotype associated with said second allele, and wherein said GCG repeat of said second allele has the sequence selected from the group consisting of:
- a) SEQ ID NO:4, when  $n=2$ ;
  - b) SEQ ID NO:5, when  $n=3$ ;
  - c) SEQ ID NO:6, when  $n=4$ ;
  - d) SEQ ID NO:7, when  $n=5$ ;
  - e) SEQ ID NO:8, when  $n=6$ ; and
  - f) SEQ ID NO:9, when  $n=7$ .